

We Claim:

1. An isolated nucleic acid molecule containing a sequence encoding a protein tyrosine phosphatase which is associated with Lafora's disease.
2. A nucleic acid according to claim 1 having a sequence as shown in SEQ.ID.NO.:1
5 or Figure 13.
3. An isolated nucleic acid molecule according to claim 1 comprising
 - (a) a nucleic acid sequence as shown in SEQ.ID.NO.:1 or Figure 13, wherein T can also be U;
 - (b) nucleic acid sequences complementary to (a);
 - 10 (c) nucleic acid sequences which are homologous to (a) or (b);
 - (d) a fragment of (a) to (c) that is at least 15 bases, preferably 20 to 30 bases, and which will hybridize to (a) to (d) under stringent hybridization conditions; or
 - (e) a nucleic acid molecule differing from any of the nucleic acids of (a) to (c) in codon sequences due to the degeneracy of the genetic code.
- 15 4. An isolated nucleic acid molecule according to claim 1 having a sequence as shown in Figure 4A.
5. An isolated nucleic acid molecule according to claim 1 having a sequence as shown in Figure 7.
6. An isolated nucleic acid molecule according to claim 1 having a sequence as
20 shown in Figure 9.
7. A method of detecting Lafora's disease comprising detecting a mutation or deletion in a nucleic acid sequence according to ~~any one of claims 1 to 6~~ in a sample from an animal.
8. A method according to claim 7 comprising detecting a mutation or deletion in a
25 region of the nucleic sequence between markers DS61003 and DS61042.
9. A method according to claim 7 comprising detecting a C to T change in nucleotide number 721 of the sequence shown in SEQ.ID.NO.:1 or Figure 13.

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10. A method according to claim 9 wherein the C to T change is detected by a method comprising:
- (a) amplifying the nucleic acid sequences in the sample with primers H1F (5'-GAATGCTCTTTCCACTTTGC-3') and PTPR (5'-GGCTCCTTAGGGAAATCAG-3') in a polymerase chain reaction;
- (b) digesting the amplified sequences with the restriction endonuclease *HaeIII*; and
- (c) determining the size of the digested sequences wherein the presence of a fragment of approximately 199bp indicates the sample is from an animal with Lafora's disease or an animal that is a carrier of Lafora's disease.

11. A method according to claim 7 comprising detecting a G to A mutation of nucleotide number 836 of the sequence shown in SEQ.ID.NO.:1 or Figure 13.

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12. A method according to claim 11 wherein the G to A change is detected by a method comprising:
- (a) amplifying the nucleic acid sequences in the sample with primers H1F (5'-GAATGCTCTTTCCACTTTGC-3') and PTPR (5'-GGCTCCTTAGGGAAATCAG-3') in a polymerase chain reaction;
- (b) digesting the amplified sequences with the restriction endonuclease *PstI*; and
- (c) determining the size of the digested sequences wherein the presence of at least one fragment of approximately 520bp indicates that the sample is from an animal that does not have Lafora's disease or an animal that is a carrier of Lafora's disease.

13. A method according to claim 7 comprising detecting a deletion of 75 kb in the sequence of EPM2A shown in SEQ.ID.NO.:1 or Figure 4A.

14. A method according to claim 7 comprising detecting a deletion of 25 kb in the sequence of EPM2A shown in SEQ.ID.NO.:1 or Figure 4A.

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15. A method of according to claim 13 or 14 comprising:
- (a) amplifying the nucleic acid sequences in the sample with primers JRGXBF (5'-TCCATTGTGCTAATGCTATCTC-3') and JRGXBR (5'-TCAGCTTGCTTTGAGGATATTT-3') in a polymerase chain reaction; and
- (b) detecting amplified sequence wherein the absence of an amplified sequence indicates that the sample is from an animal with Lafora's disease.

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16. A method according to claim 7 comprising detecting a mutation or deletion as specified in Table 3 in a sample from an animal.

17. An isolated protein containing a tyrosine phosphatase domain which is associated with Lafora's disease.

5 18. A protein according to claim 17 having the amino acid sequence as shown in SEQ.ID.NO.:2 or Figure 14.

A 19. A method for detecting Lafora's disease comprising detecting a deletion or mutation in a protein according to ~~any one of claims 17 or 18.~~ ¹⁷

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